General

Guideline Title

Use of array genomic hybridization technology in prenatal diagnosis in Canada.

Bibliographic Source(s)

Duncan A, Langlois S, SOGC Genetics Committee, CCMG Prenatal Diagnosis Committee. Use of array genomic hybridization technology in prenatal diagnosis in Canada. J Obstet Gynaecol Can. 2011 Dec;33(12):1256-9. [18 references] PubMed

Guideline Status

This is the current release of the guideline.

Recommendations

Major Recommendations

The quality of evidence (I-III) and classification of recommendations (A-L) are defined at the end of the "Major Recommendations."

Technical and Interpretive Considerations of Array Genomic Hybridization Technology

1. Array genomic hybridization is not recommended in pregnancies at low risk for a structural chromosomal abnormality; for example, advanced maternal age, positive maternal serum screen, previous trisomy, or the presence of "soft markers" on fetal ultrasound. (III-D)
2. Array genomic hybridization may be an appropriate diagnostic test in cases with fetal structural abnormalities detected on ultrasound or fetal magnetic resonance imaging; it could be done in lieu of a karyotype if rapid aneuploidy screening is negative and an appropriate turnaround time for results is assured. (II-2A)
3. Any pregnant woman who qualifies for microarray genomic hybridization testing should be seen in consultation by a medical geneticist before testing so that the benefits, limitations, and possible outcomes of the analysis can be discussed in detail. The difficulties of interpreting some copy number variants should also be discussed. This will allow couples to make an informed decision about whether or not they wish to pursue such prenatal testing. (III-A)

Definitions:

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly randomized controlled trial.

II-1: Evidence from well-designed controlled trials without randomization.

II-2: Evidence from well–designed cohort (prospective or retrospective) or case–control studies, preferably from more than one centre or research

group.

II-3: Evidence obtained from comparisons between times or places with or without the intervention. Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees.

Classification of Recommendations†

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommend the clinical preventive action.

C. The existing evidence is conflicting and does not allow to make a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

L. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

*Adapted from The Evaluation of Evidence criteria described in the Canadian Task Force on Preventive Health Care.

†Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on Preventive Health Care.

Clinical Algorithm(s)

None provided

Scope

Disease/Condition(s)

Fetal chromosomal abnormality

Guideline Category

Counseling

Diagnosis

Evaluation

Risk Assessment

Technology Assessment

Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Obstetrics and Gynecology

Intended Users

Advanced Practice Nurses

Clinical Laboratory Personnel

Nurses

Physician Assistants

Physicians

Guideline Objective(s)

To summarize for obstetrical care providers the current literature on array genomic hybridization in prenatal diagnosis and to outline the recommendations of the Canadian College of Medical Geneticists (CCMG) regarding the use of this new technology with respect to prenatal diagnosis

Target Population

Pregnant women at risk for fetal structural abnormalities

Interventions and Practices Considered

1. Array genomic hybridization testing
2. Consultation by a medical geneticist before testing on the benefits, limitations, and possible outcomes of the analysis

Major Outcomes Considered

- Detection of copy number anomalies
- Risk of unclear array genomic hybridization findings

Methodology

Methods Used to Collect/Select the Evidence

Searches of Electronic Databases

Searches of Unpublished Data

Description of Methods Used to Collect/Select the Evidence

PubMed and Medline were searched for articles published in English between 2004 and 2010, using the key words DNA QF-PCR, quantitative fluorescent polymerase chain reaction, fetal chromosomal abnormalities, prenatal diagnosis, array genomic hybridization, fetal structural anomalies, and copy number variants. Results were restricted to systematic reviews, randomized control trials/controlled clinical trials, and observational studies. Searches were updated on a regular basis, and articles were incorporated in the guideline to September 2011. Grey (unpublished) literature was identified through searching the websites of health technology assessment and health technology assessment-related agencies, clinical practice guideline collections, clinical trial registries, and national and international medical specialty societies.

Number of Source Documents

Not stated

Methods Used to Assess the Quality and Strength of the Evidence

Weighting According to a Rating Scheme (Scheme Given)

Rating Scheme for the Strength of the Evidence

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly randomized controlled trial

II-1: Evidence from well-designed controlled trials without randomization

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II-3: Evidence obtained from comparisons between times or places with or without the intervention. Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees

*Adapted from The Evaluation of Evidence criteria described in the Canadian Task Force on Preventive Health Care.

Methods Used to Analyze the Evidence

Systematic Review

Description of the Methods Used to Analyze the Evidence

The quality of evidence in this document was rated using the criteria described in the Report of the Canadian Task Force on Preventive Health Care (see the "Rating Scheme for the Strength of the Evidence" and the "Rating Scheme for the Strength of the Recommendations" fields).

Methods Used to Formulate the Recommendations

Expert Consensus

Description of Methods Used to Formulate the Recommendations

Not stated

Rating Scheme for the Strength of the Recommendations

Classification of Recommendations†

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommend the clinical preventive action.

C. The existing evidence is conflicting and does not allow to make a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

L. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

†Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on Preventive Health Care.

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Internal Peer Review

Description of Method of Guideline Validation

This technical update has been prepared by the Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Prenatal Diagnosis Committee of the of the Canadian College of Medical Geneticists (CCMG) and approved by the Executive of the SOGC.

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of supporting evidence is identified and graded for each recommendation (see the "Major Recommendations" field).

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

- Appropriate use of array genomic hybridization technology in prenatal diagnosis
- The primary advantage of array genomic hybridization is increased detection of copy number anomalies: the deviations that can be measured by molecular means are orders of magnitude smaller than those detectable by light microscopy.
- Array genomic hybridization is superior in the detection of copy number anomalies, finding a pathogenic abnormality in up to 16% of fetuses with an abnormal ultrasound and normal karyotype (see Table 2 in the original guideline document).

Potential Harms

Not stated

Qualifying Statements

Qualifying Statements

This document reflects emerging clinical and scientific advances on the date issued, and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed. Local institutions can dictate amendments to these opinions. They should be well documented if modified at the local level. None of these contents may be reproduced in any form without prior written permission of the Society of Obstetricians and Gynaecologists of Canada (SOGC).

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Foreign Language Translations

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

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Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2011 Dec

Guideline Developer(s)

Canadian College of Medical Geneticists - Professional Association

Society of Obstetricians and Gynaecologists of Canada - Medical Specialty Society

Source(s) of Funding

Society of Obstetricians and Gynaecologists of Canada (SOGC)

Guideline Committee

Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Prenatal Diagnosis Committee of the Canadian College of Medical Geneticists (CCMG)

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Financial Disclosures/Conflicts of Interest

Disclosure statements have been received from all members of the committees.

Guideline Status

This is the current release of the guideline.

Guideline Availability

Electronic copies: Available in Portable Document Format (PDF) from the Society of Obstetricians and Gynaecologists of Canada (SOGC) Web site. Also available in French from the SOGC Web site.

Print copies: Available from the Society of Obstetricians and Gynaecologists of Canada, La société des obstétriciens et gynécologues du Canada (SOGC) 780 promenade Echo Drive Ottawa, ON K1S 5R7 (Canada); Phone: 1-800-561-2416.

Availability of Companion Documents

None available

Patient Resources

None available

NGC Status

This NGC summary was completed by ECRI Institute on April 11, 2012. The information was verified by the guideline developer on May 10, 2012.

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